

Terms of Reference

1. Background

Neuromuscular disorders (NMD) cover a range of disorders including Duchenne and Becker muscular dystrophy, myotonic dystrophy, spinal muscular atrophies and other neuropathies. These disorders are associated with a range of symptoms from muscle weakness or atrophy to pain or numbness and abnormal muscle movement that can become progressively more severe. The disorders may involve difficulty with swallowing, speech or breathing and other organ systems such as the cardiac or urinary systems.¹

A survey of the literature in the Netherlands estimated the overall prevalence of neuromuscular disorders (including rare forms) in both males and females at approximately 1 in 3500². Although this is relatively uncommon the impact of NMD is enormous with the burden of disease being grossly disproportionate to its frequency.

Advances in scientific knowledge have the potential to greatly benefit patients, their families, and the community. In Australia, despite several centres being involved in research on, and management of, individuals with NMD, specifically Duchenne muscular dystrophy (DMD), there was insufficient coordination to ensure the translation of new knowledge into improved patient outcomes. Diagnosis, treatment and prevention will benefit from a more integrated approach to improve outcomes, increase innovation and decrease costs. New RNA therapeutics being developed such as the exon skipping technology offer the promise of treatment for many NMD patients. National registries have developed as a key tool to enable patients to access international clinical trials utilising the latest technology. Many countries, facing the same issues as Australia have formed a global network of national registries for NMD under the TREAT-NMD alliance.

Advocacy for a Duchenne muscular dystrophy Registry by the Duchenne Foundation and later the Muscular Dystrophy Foundation led to a request by the Australian Health Ministers Advisory Council for a report on a DMD Registry. The successful establishment of the DMD Registry has highlighted the need for an overarching Neuromuscular Disorders Registry for Australia including registries for other neuromuscular disorders.

2. Purpose

The Australian Neuromuscular Disorders Registry (ANMDR) Advisory Committee was formed to enable a range of stakeholders to facilitate the development of and provide advice on a national neuromuscular disorders registry for Australia.

¹ Amato AA and Russell JA, 2008. Neuromuscular disorders. McGraw-Hill Medical

² Emery AE, 1991. Population frequencies of inherited neuromuscular diseases – a world survey. Neuromuscular disorders Vol 1, No 1, 19 – 29.

Australian Neuromuscular Disorders Registry Advisory Committee

The specific objectives of the ANMDR Advisory Committee will be:

- To provide clinical, technical, and patient support expertise to inform the development of patient registries for neuromuscular disorders;
- To facilitate the implementation of the registries in all jurisdictions in Australia and New Zealand;
- To provide advice and share expertise on the management of neuromuscular registries in Australia and New Zealand;
- To assist with the provision of aggregate registrant information to TREAT-NMD; and
- To consider and approve applications to use the data held within the ANMDR.

3. Aim

The aim of the ANMDR Advisory Committee (The Committee) is to inform and facilitate the development of voluntary patient registries for people living with neuromuscular disorders in Australia and New Zealand.

4. Governance

The Committee will elect members ensuring representation from: each jurisdiction; paediatric and adult clinicians; patient support organisations; patients; and areas of expertise as required.

Members of The Committee will be invited up to a maximum of 20 people.

At the discretion of The Committee, sub-committees and working parties may be established for finite periods and specific tasks/activities. The size and constitution of the membership of these groups would be determined by their operational requirements.

The Office of Population Health Genomics will provide: an Executive Chair; executive support; and secretariat duties for The Committee.

The Committee will evaluate and report on its activities annually. The report will be made available to the public on the website.

(<http://www.nmdregistry.com.au/>)

5. Membership

The Committee membership will reflect the diversity of people involved in clinical and diagnostic services as well as members of the community who are living with neuromuscular disorders. The Committee will have the authority to co-opt other members as required or establish working parties for specific approved projects.

The Committee is committed to supporting and enabling equal opportunities and participation for all Australian's living with neuromuscular disorders and will consider the specific needs of particular sectors in our community, including Aboriginal people, people from linguistically diverse backgrounds, people with disabilities and those people living in regional and remote locations.

Australian Neuromuscular Disorders Registry Advisory Committee

The Chair of the Committee will undertake an executive role and be responsible for leading the development and implementation of neuromuscular registries.

Selection of Committee Members

New appointments to The Committee will be made by nomination and Committee voting. A unanimous Committee decision is required and the Chair is responsible for extending the invitation and providing the new member with the following:

- A copy of the Charter and Terms of Reference;
- A declaration to act as a member of The Committee; and
- A disclosure of potential conflicts of interest in their duties as a member of The Committee.

6. Term

The Committee role is set at 4 years from June 2012.

To enable The Committee to have continuity of membership, initial appointment terms will be staggered with half of the committee members appointed for two years, and half appointed for three years. Retiring members may apply for re-appointment for a second term.

The Committee will set a review period for the ongoing operational role and coordinating functions of The Committee at the anniversary date of the first Committee meeting.

7. Meetings

The ANMDR Advisory Committee will:

- Meet four to six weekly;
- Review the frequency of meetings as required by the work flow;
- Limit the duration of the meetings to one hour; and
- Determine the meeting times as convenient for the sitting members.

8. Member Obligations

The ANMDR Advisory Committee members will:

- Pre-read the meeting documents;
- Be consulted on an *ad-hoc* basis by the chairman between meetings if required; and
- Arrange proxies to attend/participate in the place of committee members advising the Executive Chair/Secretariat in writing one week prior to the scheduled meeting.

Absence from three (3) consecutive meetings, without prior arrangements made with the Chair/Secretariat will be grounds for retiring a member and appointing a replacement for the Committee vacancy.

9. Quorum

A quorum shall consist of half the committee membership plus one including the Chair. In the absence of a quorum, any discussion or decisions will require

ratification by The Committee members either via circular resolution or will be held over for discussion and voting at the next meeting.

10. Support

All paperwork relating to The Committee meetings will be provided in an appropriate and timely manner, usually at least one week prior to The Committee meeting.

11. Terms of Reference

The ANMDR Advisory Committee members will:

- Provide clinical, technical and patient expertise to enable the development of neuromuscular registries appropriate for Australian patients;
- Facilitate the implementation of the neuromuscular registries in their respective jurisdictions;
- Coordinate and assist with the addition of clinical and molecular data to the neuromuscular registries in their respective jurisdictions;
- Review and provide advice on applications received for access to the data held in the NMDR ; and
- Assist with establishing links with and providing information to neuromuscular stakeholders.

The ANMDR Advisory Committee Terms of Reference will be reviewed annually.

Revision History:

Draft 1.0 by Leanne Youngs, 23 August 2012.

Endorsement:

Australian Neuromuscular Disorders Registry Advisory Committee at teleconference Tuesday July 17th 2012.

12. Appendix 1: Australian Neuromuscular Disorders Registry Advisory Committee Membership

Dr Hugh Dawkins (Chair, National Curator)	Office of Population Health Genomics, Department of Health
Dr Anita Cairns (QLD)	Royal Children's Hospital & Mater Children's Hospital Brisbane
Dr Damian Clark (SA)	Women's and Children's Hospital Adelaide
Dr Alastair Corbett (NSW)	Concord Repatriation General Hospital
Mark Davis (WA)	Neurogenetics Unit, Royal Perth Hospital
Desiree Du Sart (VIC)	Murdoch Institute, Royal Children's Hospital
Dr Michelle Farrar (NSW)	Neurology Department, Sydney Children's Hospital
John Gummer (WA)	Muscular Dystrophy Foundation
Dr Emma Hammond	Office of Population Health Genomics, Department of Health
Dr Graeme Hammond-Tooke (NZ)	
Dr Val Hyland (QLD)	Molecular Genetic Unit, Royal Brisbane Hospital
Dr Alexa Kidd (NZ)	
Dr Phillipa Lamont (WA)	Neurology, Royal Perth Hospital
Lesley Murphy (WA)	Parent
Dr Kathryn North (NSW)	Institute for Neuromuscular Research, Children's Hospital at Westmead
Deborah Robins (QLD)	Duchenne Foundation
Miriam Rodrigues (NZ)	Muscular Dystrophy Association New Zealand
Dr Peter Rowe (WA)	Neurology, Princess Margaret Hospital for Children
Dr Monique Ryan (VIC)	Neurosciences Department, The Royal Children's Hospital Melbourne
Boris Struk (VIC)	Muscular Dystrophy Association
Dr Peter Taylor (NSW)	South Eastern Area Laboratory Services, Prince of Wales Hospital
Dr Sui Yu (SA)	Department of Genetic Medicine, Women's and Children's Hospital Adelaide